

Letter to the Editor

Porter et al (2001) should be commended for their insightful report on monozygotic (MZ) female twins concordant for intracranial aneurysms¹.

Following detection of this condition in one twin, the co-twin was similarly diagnosed and both twins were treated successfully. This was only the tenth such case to appear in the literature, and so directs attention to a "rare and unique subtype" of familial aneurysm. Only one of the previous studies described discordance in an MZ twin pair. The authors are, thus, correct in urging reports of discordant cases to determine the risk of aneurysm to MZ co-twins of affected twins, and to twins in general.

It is unfortunate that Porter et al did not document the method by which the zygosity of the pair in question was determined. This is best accomplished via serological analyses or molecular genetic studies². Failure to do so may raise questions surrounding interpretation of the findings. Twins in this study showed differences in presentation and anatomy, mandating different treatment plans. Such discrepancies are clearly intriguing in MZ twins, but in the absence of zygosity validation the possibility that the pair could be dizygotic (DZ) cannot be dismissed. Other investigators may hesitate to include this case in pooled analyses.

Twins in this report were referred to as "identical". Currently, there is preference among twin researchers for replacing the familiar label "identical" with the scientific term "monozygotic"³. Identical implies perfect co-twin concordance in be-

havior and physique which is known to be incorrect. Even while MZ twins show greater resemblance across most measured traits than other pairs of relatives (demonstrating genetic influence), they do not show perfect similarity. Sources of MZ co-twin discordance include post-zygotic genetic events (e.g., chromosomal mosaicism), mirror-image reversal in mostly ectodermally derived traits (e.g., dermatoglyphics), midline neurological malformations (e.g., spina bifida) and unequal fetal blood supply (e.g., fetal anastomoses)⁴. The fact the twins in the present report did display presentation and anatomical differences is, therefore, not inconsistent with a diagnosis of monozygosity? But knowing their twin type with certainty allows their differences and similarities appropriate scientific scrutiny.

References

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- 2 Segal NL: *Entwined lives: Twins and what they tell us about human behavior*. NY: Plume, 2000.
- 3 Segal NL: Identical and fraternal: When words mislead. *Twin Research* 3: 383-343, 2000.
- 4 Machin GA: Some causes of genotypic and phenotypic discordance in monozygotic twin pairs. *Am J Hum Genet* 61: 216-228, 1996.

Nancy L. Segal, M.D.
Psychology Department
California State University, Fullerton
800 N. State College Blvd.
Fullerton, CA 92834, USA
nsegal@fullerton.edu

Response

The authors are grateful to Dr. Segal for her insightful comments and recommendations of more precise terminology for our case description.

These twins have always been told they are "identical" by their physicians in the past, but unfortunately their zygosity has never been formally verified. There is a history of twin births on the maternal and paternal sides. Discordant features in monozygotic twins provide evidence for the importance of the "fourth dimension", time, in the evolution of pathological states of the cerebral blood vessels¹.

As clinicians, we tend to be faced with a symptomatic patient experiencing an acute problem related to the vascular system, and may lose perspective on the effect of time on the disease. As Dr. Segal points out, post-zygotic genetic (and also environmental) events may lead to differences in anatomy or symptomatology in monozygous patients who thus are not truly "identical". The case we present is interesting for both the similarities and the differences demonstrated in the vessels of these patients.

Dr. Segal's comments have reminded us that enlisting the opinions of experts outside of our own narrow discipline often enhances the learning experience for a unique case like ours.

References

- 1 Lasjaunias PL: Segmental identity and vulnerability in cerebral arteries. *Interventional Neuroradiology* 6: 113-124, 2000.

Phillip J. Porter M.D., F.R.C.S.(C)
Neurosurgery & Interventional Neuroradiology
Toronto Western Hospital, UHN
phillip.porter@uhn.on.ca